



# TRIS project: An overview of the database and project activities

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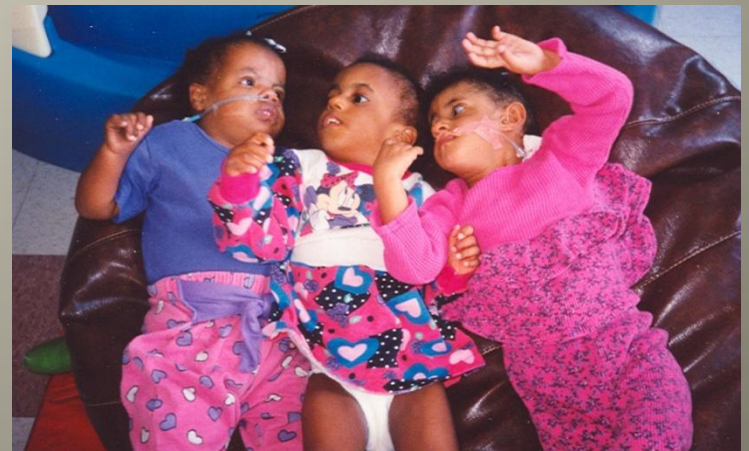


# Tracking Rare Incidence Syndromes

The Tracking Rare Incidence Syndromes (TRIS) project seeks to increase awareness and knowledge for families and professionals touched by rare trisomy conditions and aims to facilitate improved decision making for optimal services and supports for affected children and their families.

# Origins of the TRIS project

- ❖ Principal Investigator's experience with young children with full trisomy 18 in early 1990's
- ❖ Threads on Tri-family and Tri-med lists including prenatal issues, family needs, working with professionals & medical concerns
- ❖ TRIS was “born” in 2003; Advisory Committee and a planning group formed; pilot data collected in 2005-06; online TRIS Survey launched 2/1/07



# TRIS project components

- ❖ Database development through parent-to-parent contact, links with rare trisomy groups and online searches
- ❖ TRIS Survey (Full and Modified versions) includes items about birth history, medical issues, developmental progress and support needs.  
In addition, annual TRIS Follow-up Survey for updates in key areas.
- ❖ Dissemination through presentations and publications
- ❖ Collaboration with experts in field
- ❖ Outreach for raising awareness and recruitment

# TRIS Survey

- Topics include:
  - ❖ Pregnancy and postnatal period
  - ❖ Long-term survival data
  - ❖ Family support
  - ❖ Feeding needs
  - ❖ Medical interventions including surgery
  - ❖ Developmental milestones



# TRIS project database

- Approximately 400 children represented in TRIS project database
- TRIS Survey Completions
  - Full Survey ( $> 2$  months) = 197
  - Modified Survey ( $< 2$  months) = 146
  - Follow-up Survey (annual)
    - ❖ Year 1 = 112
    - ❖ Year 2 = 64
    - ❖ Year 3 = 22
    - ❖ Year 4 = 5





# Dissemination: Presentations

- ❖ Support Organization for Children with Trisomy 18, 13 and Related Disorders Conference (2006 to present)
- ❖ American Academy for Cerebral Palsy and Developmental Medicine (2008)
- ❖ Council for Exceptional Children Convention (2010)
- ❖ Division for Early Childhood Conference (2006, 2007, 2009)
- ❖ National Advanced Practice Neonatal Nurses Conference (2008)



# Dissemination:

## Publications

Bruns, D. A. (2008). Pregnancy and birth history of newborns with Trisomy 18 or 13: A pilot study. *American Journal of Medical Genetics Part A*, 146A(3), 321-326.

- ❖ Presented initial TRIS Survey data (n=27); greater than 10% one year survival rate (longer for children with t13 subtypes)
- ❖ Overview of birth data, medical issues and newborn interventions





# Dissemination:

## Publications continued



Bruns, D. A. (2010). Neonatal experiences of newborns with full Trisomy 18. *Advances in Neonatal Care*, 10(1), 25-31.

- ❖ 21 newborns with full t18 (13 survivors; 8 who died prior to mothers' participation in the TRIS project); gestational age was generally older than described in the literature
- ❖ Newborns presented with syndrome-related physical characteristics (e.g., low-set ears, small jaw) and medical conditions (e.g., heart defects)
- ❖ In NICU, newborns were provided with respiratory interventions (e.g., nasal canula, pulse oximeter) and received treatments including blood transfusions and echocardiograms.
- ❖ Data also indicate longer survival rates than present in the literature ( $M=103.3$  months for survivors, 37.5 months for angels)

# Dissemination: Publications continued



Bruns, D. & Foerster, K. (2011).

“We’ve been through it all together: Supports for parents with children with rare trisomy conditions. *Journal of International Disability Research*, 55(4), 361-369.

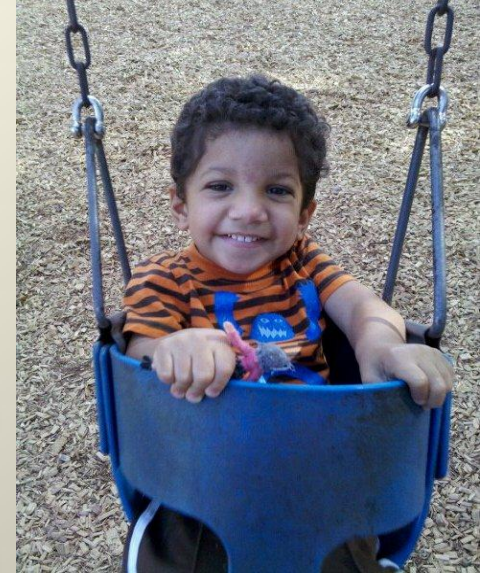
- ❖ Support from immediate and extended family members varied from very positive to participants describing very negative interactions with specific individuals; prognosis affected availability of support
- ❖ Many in the sample (n=20 total, n=6 trisomy 9, n=7 trisomy 13, n=7 trisomy 18) reported affirming experiences with spouses and difficulties with grandparents and other extended family members.

# Dissemination:

## Publications continued

Bruns, D. A. (in press). Presenting physical characteristics, medical conditions, and developmental status of long-term survivors with trisomy 9 mosaicism. *American Journal of Medical Genetics Part A*.

- ❖ Fourteen long-term survivors ( $M=78.6$  months, range = 2 - 293 months,  $SD = 89.32$  months; 3 – 99% mosaic); results indicated the presence of low set ears and microcephaly for some children in the sample
- ❖ Cardiac anomalies were reported along with feeding and respiratory difficulties in the immediate postnatal period
- ❖ Data from TRIS Developmental Matrix indicated a wide range in functioning level

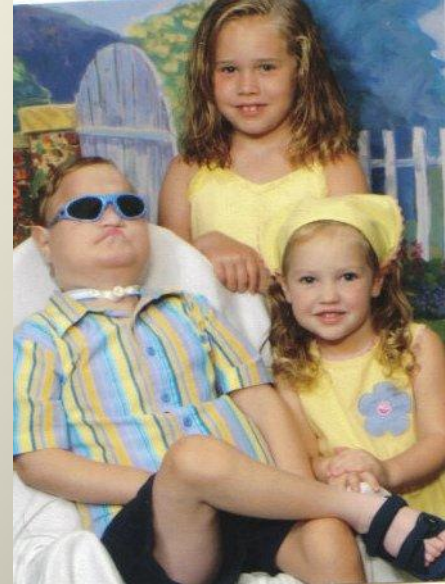


# Dissemination:

## Publications continued

Bruns, D. (in review). Physical characteristics and presenting medical conditions in long-term survivors with full trisomy 13. Submitted to *American Journal of Medical Genetics Part A*.

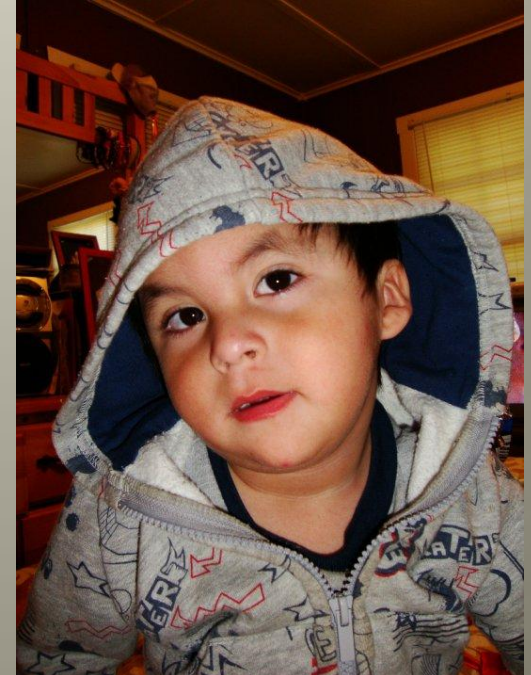
- ❖ Longer mean survival rates:  $M = 48.4$  months survivors living at time of data collection and 40.8 months for those who died prior
- ❖ Presence of common physical characteristics (e.g., low-set ears, cleft palate) and medical conditions (e.g., cardiac defects, feeding difficulties)





# Implications

- Raise awareness of Early Intervention professionals about this population
- Need to re-examine existing literature based on TRIS Survey results
- Provide support to families
- Continue growing TRIS project database; broaden international scope with translations  
(Modified Survey underway in Spanish)



# Collaboration

- Medical professionals in general medicine, genetics and bioethics
- Parents with living children (e.g., mother of daughter with full trisomy 18 (28 years) and President of the Support Organization for 18, 13 and Related Disorders) as well as parents with children who have died (e.g., mother of daughter with full trisomy 13 (angel) and advocate)





# Outreach

- Continue work with collaboration partners
- Preparation is underway of informational DVD
- Social media outlets such as rare trisomy groups on Facebook
- Parent to parent contacts



# Future plans

- Continue analysis of TRIS Survey data
- Gain additional funding for systematic outreach efforts
- Present in additional venues
- Continue to prepare publications

